



FRMD7 gene

FERM domain containing 7

Normal Function

The *FRMD7* gene provides instructions for making a protein whose exact function is unknown. This protein is found in many tissues, but it is most abundant in areas of the brain that control eye movement (such as the midbrain and cerebellum) and in the light-sensitive tissue at the back of the eye (retina). The FRMD7 protein likely plays a role in the development of nerve cells in these areas of the brain and the retina.

Health Conditions Related to Genetic Changes

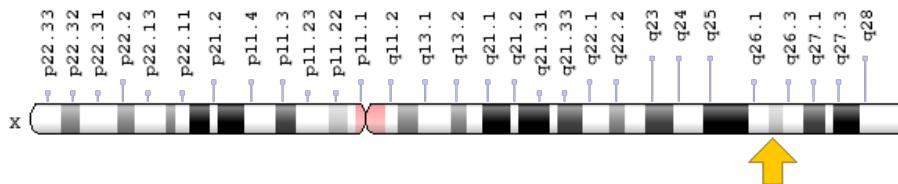
X-linked infantile nystagmus

More than 35 mutations in the *FRMD7* gene have been found to cause X-linked infantile nystagmus. Most of these mutations change single protein building blocks (amino acids) in the FRMD7 protein. Mutations in the *FRMD7* gene likely lead to the production of a protein that is unstable and that cannot perform its normal function. A lack of functional FRMD7 protein is thought to disrupt the development of nerve cells in the retina and areas of the brain that control eye movement. Abnormal development of these nerve cells likely causes the involuntary side-to-side eye movements that are characteristic of X-linked infantile nystagmus.

Chromosomal Location

Cytogenetic Location: Xq26.2, which is the long (q) arm of the X chromosome at position 26.2

Molecular Location: base pairs 132,076,986 to 132,128,022 on the X chromosome (Homo sapiens Annotation Release 108, GRCh38.p7) (NCBI)



Credit: Genome Decoration Page/NCBI

Other Names for This Gene

- FRMD7_HUMAN
- NYS1

Additional Information & Resources

GeneReviews

- FRMD7-Related Infantile Nystagmus
<https://www.ncbi.nlm.nih.gov/books/NBK3822>

Scientific Articles on PubMed

- PubMed
<https://www.ncbi.nlm.nih.gov/pubmed?term=%28FRMD7%5BTIAB%5D%29+AND+%28Genes%5BMH%5D%29+OR+%28Genetic+Phenomena%5BMH%5D%29%29+AND+english%5Bla%5D+AND+human%5Bmh%5D+AND+%22last+3600+days%22%5Bdp%5D>

OMIM

- FERM DOMAIN-CONTAINING PROTEIN 7
<http://omim.org/entry/300628>

Research Resources

- Atlas of Genetics and Cytogenetics in Oncology and Haematology
http://atlasgeneticsoncology.org/Genes/GC_FRMD7.html
- ClinVar
<https://www.ncbi.nlm.nih.gov/clinvar?term=FRMD7%5Bgene%5D>
- HGNC Gene Family: FERM domain containing
<http://www.genenames.org/cgi-bin/genefamilies/set/1293>
- HGNC Gene Symbol Report
http://www.genenames.org/cgi-bin/gene_symbol_report?q=data/hgnc_data.php&hgnc_id=8079
- NCBI Gene
<https://www.ncbi.nlm.nih.gov/gene/90167>
- UniProt
<http://www.uniprot.org/uniprot/Q6ZUT3>

Sources for This Summary

- OMIM: FERM DOMAIN-CONTAINING PROTEIN 7
<http://omim.org/entry/300628>
- He X, Gu F, Wang Z, Wang C, Tong Y, Wang Y, Yang J, Liu W, Zhang M, Ma X. A novel frameshift mutation in FRMD7 causing X-linked idiopathic congenital nystagmus. *Genet Test.* 2008 Dec;12(4):607-13. doi: 10.1089/gte.2008.0070.
Citation on PubMed: <https://www.ncbi.nlm.nih.gov/pubmed/19072571>
- Li N, Wang L, Cui L, Zhang L, Dai S, Li H, Chen X, Zhu L, Hejtmancik JF, Zhao K. Five novel mutations of the FRMD7 gene in Chinese families with X-linked infantile nystagmus. *Mol Vis.* 2008 Apr 18;14:733-8.
Citation on PubMed: <https://www.ncbi.nlm.nih.gov/pubmed/18431453>
Free article on PubMed Central: <https://www.ncbi.nlm.nih.gov/pmc/articles/PMC2324116/>
- Self JE, Shawkat F, Malpas CT, Thomas NS, Harris CM, Hodgkins PR, Chen X, Trump D, Lotery AJ. Allelic variation of the FRMD7 gene in congenital idiopathic nystagmus. *Arch Ophthalmol.* 2007 Sep;125(9):1255-63.
Citation on PubMed: <https://www.ncbi.nlm.nih.gov/pubmed/17846367>
- Shiels A, Bennett TM, Prince JB, Tychsen L. X-linked idiopathic infantile nystagmus associated with a missense mutation in FRMD7. *Mol Vis.* 2007 Nov 29;13:2233-41.
Citation on PubMed: <https://www.ncbi.nlm.nih.gov/pubmed/18087240>
- Tarpey P, Thomas S, Sarvananthan N, Mallya U, Lisgo S, Talbot CJ, Roberts EO, Awan M, Surendran M, McLean RJ, Reinecke RD, Langmann A, Lindner S, Koch M, Jain S, Woodruff G, Gale RP, Bastawrous A, Degg C, Droutsas K, Asproudis I, Zubcov AA, Pieh C, Veal CD, Machado RD, Backhouse OC, Baumber L, Constantinescu CS, Brodsky MC, Hunter DG, Hertle RW, Read RJ, Edkins S, O'Meara S, Parker A, Stevens C, Teague J, Wooster R, Futreal PA, Trembath RC, Stratton MR, Raymond FL, Gottlob I. Mutations in FRMD7, a newly identified member of the FERM family, cause X-linked idiopathic congenital nystagmus. *Nat Genet.* 2006 Nov;38(11):1242-4. Epub 2006 Oct 1. Erratum in: *Nat Genet.* 2011 Jul;43(7):720. Bastawrous, Andrew [added].
Citation on PubMed: <https://www.ncbi.nlm.nih.gov/pubmed/17013395>
Free article on PubMed Central: <https://www.ncbi.nlm.nih.gov/pmc/articles/PMC2592600/>
- Zhang B, Liu Z, Zhao G, Xie X, Yin X, Hu Z, Xu S, Li Q, Song F, Tian J, Luo W, Ding M, Yin J, Xia K, Xia J. Novel mutations of the FRMD7 gene in X-linked congenital motor nystagmus. *Mol Vis.* 2007 Sep 13;13:1674-9.
Citation on PubMed: <https://www.ncbi.nlm.nih.gov/pubmed/17893669>
- Zhang Q, Xiao X, Li S, Guo X. FRMD7 mutations in Chinese families with X-linked congenital motor nystagmus. *Mol Vis.* 2007 Aug 3;13:1375-8.
Citation on PubMed: <https://www.ncbi.nlm.nih.gov/pubmed/17768376>

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